

CLAIMS

We claim:

1. A method for characterizing a nucleic acid region, the method comprising
 - (a) adding to each of a plurality of reaction chambers a nucleic acid sample and a different set of amplification primers, wherein each set of amplification primers is complementary to a single amplicon of a nucleic acid region of interest;
 - (b) performing amplification reactions for each reaction chamber under the same reaction conditions;
 - (c) bringing into contact in each of a plurality of reaction chambers an amplicon from a different one of the amplification reactions and one or more internal sequencing primers corresponding to the amplicon;
 - (d) performing sequencing reactions for each reaction chamber under the same reaction conditions; and
 - (e) analyzing the sequences of the amplicons.
2. The method of claim 1, wherein the nucleic acid region of interest is a multi-exon gene.
3. The method of claim 2, wherein the multi-exon gene is dystrophin, SOD-1 NF-1, ATM, dysferlin, calpain, sarcoglycans, collagen VI, Nebulin, or Titin.
4. The method of claim 2, wherein the amplicons collectively comprise sequence from every exon of the multi-exon gene.
5. The method of claim 4, wherein the amplicons each comprise an exonic region or proximal promoter segment of the multi-exon gene.
6. The method of claim 1, wherein at least 30 amplicons of the nucleic acid region of interest are amplified.
7. The method of claim 1, wherein a single solid support comprises all of the reaction chambers.
8. The method of claim 7, wherein the solid support is a 96 well plate.
9. The method of claim 1, wherein the amplification reactions are PCR reactions and wherein the sequencing reactions are cycle sequencing reactions.
10. The method of claim 1, wherein the amplicons produced in the amplification reactions are purified prior to step (c) and wherein the sequencing products produced in the sequencing reactions are purified prior to step (e).

11. The method of claim 1, wherein the sequences of the amplicons are analyzed by electrophoretic separation and fluorescent detection of nucleotides on a sequence analyzer.
12. The method of claim 11, wherein the sequences of the amplicons are further analyzed by identifying mutations in the nucleic acid region of interest.
13. The method of claim 12, wherein the mutations are deletions, point mutations, frameshifts, or combinations thereof.
14. The method of claim 1, wherein the sets of amplification primers are selected from the group of primer sets as shown in Table 1 or Table 6.
15. The method of claim 1, wherein the sets of sequencing primers are selected from the group of primer sets as shown in Table 2 or Table 7.
16. The method of claim 1, wherein the nucleic acid sample was derived from a patient, wherein the analysis of the sequences of the amplicons indicates dystrophinopathy in the patient.
17. The method of claim 16, wherein the dystrophinopathy is Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD).
18. The method of claim 1, wherein the sequences of the amplicons are analyzed by comparing the sequences of the amplicons to other known nucleotide sequences.
19. A primer set which recognizes a single exon or a proximal promoter for the dystrophin gene, the set comprising the primers as shown in Table 1 or Table 6.
20. A primer set which recognizes a single exon or a proximal promoter for the dystrophin gene, the set comprising the primers as shown in Table 2 or Table 7.